

# ■ Genetic diagnostic testing

## MTHFR gene test

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### Used to assess

MTHFR, or methylenetetrahydrofolatereductase, is essential in the conversion of homocysteine to methionine via methylation. A polymorphism of the gene results in defective metabolism of folate and subsequent hyperhomocysteinaemia.

### Clinical relevance

High blood levels of homocysteine are recognised as a risk factor for:

- Obesity and Type 2 Diabetes
- Coronary artery disease
- Venous thrombosis and stroke

High homocysteine levels in the blood have also been associated with:

- Migraine
- Neural tube defects
- Stillbirth
- Spontaneous/recurrent miscarriage
- Depression
- Autism spectrum disorders

### Specimen requirements

Blood sample or Buccal swab

### Specimen collection method

Healthscope Pathology collection centre for blood or self-collection test kit (mail order) for Buccal swab

*As the only Australian pathology laboratory offering the expertise of a **dedicated Genomics department**, we are proud and excited to be at the forefront of this emerging field.*

*More genetic tests will be added to our service offering in 2010-11.*